

## INFORMED CONSENT FOR HUNTINGTON DISEASE (HD) DNA TESTING

Patient Last Name \_\_\_\_\_ First Name \_\_\_\_\_ Birth Date \_\_\_\_ / \_\_\_\_ / \_\_\_\_

Does the patient have symptoms of HD? \_\_\_\_\_ If so, describe \_\_\_\_\_

Who is the patient's closest relative with HD? \_\_\_\_\_ Age the relative's symptoms began \_\_\_\_\_

Was this relative's diagnosis confirmed by DNA testing? \_\_\_\_\_

- HD is an inherited, neurodegenerative condition affecting thought processes, movement, personality, or mood. Although treatments for symptoms are available, there is currently no cure for HD. This blood test involves determining the number of CAG repeats in the HD gene. An expanded number of CAG repeats causes most cases of HD. The certainty of a positive HD DNA test result exceeds 99%. The certainty of a negative test result depends on the accuracy of the HD diagnosis in the family.
- Test results may reveal non-paternity or that other family members may be affected with, or at risk for developing, HD.
- If a gene mutation is identified, insurance rates, obtaining disability or life insurance, and employability could be affected. Federal law extends some protections regarding genetic discrimination (<http://www.genome.gov/10002328>). It is my responsibility to consider the possible impact of these results. All test results are released to the ordering health care provider and those parties entitled to them by state and local laws.
- There are four possible test results:
  - 1) **Negative:** Both of the HD genes have normal numbers of CAG repeats (<27). This individual is neither at risk for developing HD nor of passing it on to his/her offspring.
  - 2) **Intermediate:** The HD gene contains 27-35 repeats. This individual is not at risk for developing HD; however, he or she may have children with HD.
  - 3) **Affected/ Reduced Penetrance:** The HD gene contains 36-39 CAG repeats; therefore, this individual may or may not develop HD. He or she may have children with HD.
  - 4) **Affected:** The HD gene contains 40 or more CAG repeats; therefore, this individual will develop HD sometime in his/her life. Offspring of this individual have a 50% risk for developing HD. The age of symptom onset and how quickly the disease progresses cannot be precisely predicted. Neurological examination is needed to establish the onset of symptoms.
- There are psychological risks associated with HD testing. A test result predicting an uncertain clinical outcome can be frustrating. A negative result can produce feelings of guilt as well as joy. A positive result could lead to serious psychological consequences including feelings of depression, futility, despair, and severe stress. Counseling should be provided before and after testing.
- The ordering health care provider must relay the test results in person and be available for follow-up genetic counseling, as necessary. I should be accompanied by a support person, who is not at risk for HD, when receiving my result. Questions regarding result interpretation or medical management should be directed to the ordering health care provider.
- Consent may be given for allowing the sample to be used for test validation, education and stored indefinitely as long as patient privacy is maintained. Refusal to provide consent will not affect the test result. If a response is not checked, consent is implied.

I authorize consent for the use of the above named individual's sample for test validation and education.  Yes  No

**Patient or Legal Guardian section:**  
 I have the legal authority to request ARUP Laboratories to test this sample for Huntington Disease (HD). I am either the above patient or his/her legal guardian. I have been counseled regarding the risks, benefits, and limitations of knowing the test results and have carefully considered the psychological impact the results may have on the patient and his/her family.

Patient/Legal Guardian  
 Printed Name: \_\_\_\_\_ Signature \_\_\_\_\_ Date \_\_\_\_\_

**Ordering Health Care Provider or Genetic Counselor:** I have explained the above information to the patient or his/her legal guardian.

Printed Name \_\_\_\_\_ Signature \_\_\_\_\_ Practice Specialty: \_\_\_\_\_

Phone Number (\_\_\_\_) \_\_\_\_\_ FAX (\_\_\_\_) \_\_\_\_\_ Date: \_\_\_\_\_